

(19) World Intellectual Property  
Organization  
International Bureau



(43) International Publication Date  
3 February 2005 (03.02.2005)

PCT

(10) International Publication Number  
**WO 2005/010145 A3**

- (51) International Patent Classification<sup>7</sup>: **C07H 21/04**,  
C12Q 1/68
- (21) International Application Number:  
PCT/US2004/015587
- (22) International Filing Date: 9 June 2004 (09.06.2004)
- (25) Filing Language: English
- (26) Publication Language: English
- (30) Priority Data:  
60/485,301 5 July 2003 (05.07.2003) US  
60/525,859 1 December 2003 (01.12.2003) US
- (71) Applicant (for all designated States except US): **THE  
JOHNS HOPKINS UNIVERSITY** [US/US]; 3400 N.  
Charles Street, Baltimore, MD 21218 (US).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): **DRESSMAN,  
Devin** [US/US]; 701 S. Luzerne Avenue, Baltimore, MD  
21224 (US). **YAN, Hai** [CN/US]; 7236 Montgomery Road  
#3C, Elkridge, MD 21075 (US). **KINZLER, Kenneth,  
W.** [US/US]; 1403 Halkirk Way, Baltimore, MD 21015  
(US). **VOGELSTEIN, Bert** [US/US]; 3700 Breton Way,  
Baltimore, MD 21208 (US).
- (74) Agent: **KAGAN, Sarah, A.**; Banner & Witcoff, Ltd., 11th  
Floor, 1001 G Street, N.W., Washington, DC 20001-4597  
(US).
- (81) Designated States (unless otherwise indicated, for every  
kind of national protection available): AE, AG, AL, AM,  
AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN,  
CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI,  
GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE,  
KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD,  
MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG,  
PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM,  
TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM,  
ZW.
- (84) Designated States (unless otherwise indicated, for every  
kind of regional protection available): ARIPO (BW, GH,  
GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM,  
ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM),  
European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI,  
FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI,  
SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ,  
GW, ML, MR, NE, SN, TD, TG).
- Published:  
— with international search report
- (88) Date of publication of the international search report:  
11 August 2005
- For two-letter codes and other abbreviations, refer to the "Guid-  
ance Notes on Codes and Abbreviations" appearing at the begin-  
ning of each regular issue of the PCT Gazette.

(54) Title: METHOD AND COMPOSITIONS FOR DETECTION AND ENUMERATION OF GENETIC VARIATIONS

(57) Abstract: Many areas of biomedical research depend on the analysis of uncommon variations in individual genes or transcripts. Here we describe a method that can quantify such variation at a scale and ease heretofore unattainable. Each DNA molecule in a collection of such molecules is converted into a single particle to which thousands of copies of DNA identical in sequence to the original are bound. This population of beads then corresponds to a one-to-one representation of the starting DNA molecules. Variation within the original population of DNA molecules can then be simply assessed by counting fluorescently-labeled particles via flow cytometry. Millions of individual DNA molecules can be assessed in this fashion with standard laboratory equipment. Moreover, specific variants can be isolated by flow sorting and employed for further experimentation. This approach can be used for the identification and quantification of rare mutations as well as to study variations in gene sequences or transcripts in specific populations or tissues.

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